What do IOWA's "Experts" say?

A summary of interviews about the Newborn Screening Program and perspectives on adding New Conditions

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Who are the "Experts"?

Inherited Disease Specialist Pg 3

Parent Perspective Pg 5

Culture & Access to Services Pg 7

Public Transportation Pg 9

Laboratory Services Pg 11

Newborn Screening Follow-up Program Pg 13

Other States' Experiences Pg 15

Our Funder for this Event Pg 17

March of Dimes Pg 19

Local Pediatricians Pg 21

State Public Health Pg 24

Legal Considerations Pg 26

Community Economics Pg 28

Insurance Coverage Pg 30

Inherited Disease Specialist
University of Iowa

John Bernat, MD, PhD

Clinical Geneticist, Medical Director of the Iowa Lysosomal Disorders Center

"I support adding Pompe Disease and MPS-1 to Iowa's newborn screening panel as this would allow for earlier diagnosis for both of these disorders…

"While neither disease has a cure, both have treatments that can slow the progression of disease. Treatments are more effective when started earlier and give these patients the best possible chance at a positive outcome. I believe this outweighs the negative aspects of screening for these conditions, which are also important and must be carefully considered."

Who are you and what is your role in Newborn Screening?

I'm a medical consultant to the Iowa Newborn Screening Program. I treat patients with some of the conditions identified by the Newborn Screening Program. Our clinic serves the patients identified with MPS-I or Pompe Disease. We also have a medical consultant at the University of Iowa to serve patients with X-ALD.

How would adding new conditions to Iowa's Newborn Screening Panel affect your work? (Current new conditions- Pompe, MPS-1, X-ALD)

Pompe Disease and MPS-1 are both examples of Lysosomal Storage Disorders. Patients screening positive for Pompe Disease or MPS-1 would be referred to my Lysosomal Storage Disorders clinic for further evaluation. If a diagnosis is made, treatment would be started, and I would follow these patients long-term.

Making estimates based on the initial data from our neighboring states who are screening for these conditions, I would expect to receive 1-2 positive screens weekly for both disorders combined. Most of these patients would ultimately be false positives. This increase in workload is certainly manageable. In the end, Iowans deserve to benefit from screening for these conditions as well.

How would it affect the people you serve?

As with newborn screening for any disorder, people may be affected positively or negatively. Babies with the severe forms of Pompe Disease or MPS-1 can be identified and have treatment started earlier, often before they become seriously ill.

But with any screening test, false positive results can occur, causing worry in some families until other tests return negative. Milder forms of the disease can also be diagnosed, sometimes decades before the onset of any symptoms. However, these negative effects are similar for other disorders already on the newborn screening panel.

How would it affect your community?

I feel the community would become more aware of lysosomal storage disorders such as Pompe Disease and MPS-1. Although both are rare diseases, the increased diagnosis and exposure from adding them to the Iowa Newborn Screening Panel would allow for greater knowledge and resources in the community, including among local physicians, therapists, schools, and other community organizations. This would be a positive impact for patients with these rare disorders.

Do you think it's a good idea to add these conditions to the panel? Why or why not?

I support adding Pompe Disease and MPS-1 to Iowa's newborn screening panel as this would allow for earlier diagnosis for both of these disorders.

The severe form of Pompe Disease is typically diagnosed around 4 months of age, when affected babies are not gaining weight, not developing normally, and starting to have heart and breathing problems.

The severe form of MPS-1 is typically diagnosed around 9 months of age, when affected babies have bone and joint problems, growth problems, vision and hearing problems, and breathing problems.

Newborn screening would allow for a diagnosis to be made in the first few weeks to months of life, often before symptoms have started. While neither disease has a cure, both have treatments that can slow the progression of disease.

Treatments are more effective when started earlier and give these patients the best possible chance at a positive outcome. I believe this outweighs the negative aspects of screening for these conditions, which are also important and must be carefully considered.

Parent perspective

"Please, as a mother who has had two children affected by this terrible disease, I beg you to include Pompe's Disease on the newborn screening. Spare other families the hurt and loss that occurs without early notification."

-Jean Kelly
mother of two children with Pompe Disease

My name is Jean Kelly, and I would like to share with you how Pompe's Disease has affected my family's lives. We live in Pacific Junction, IA, on an acreage in the Loess Hills, which is twenty miles south of Omaha, NE. My husband and I have three sons; Austen, Ryan, and Jason. Austen, our oldest son, was born in 1992. He was a great baby, and we could not have been happier.

Our second son, Ryan, was born two years later in 1994. From the time he was born, we noticed he was not as easy of a baby as Austen. But, we were busier with work and accepted it as normal life with two children instead of one. As time wore on, we began to feel that something was not quite right. Ryan was not as active as Austen and rarely smiled. At his six month check-up, our pediatrician referred him to a pediatric neurologist, based on his low muscle tone. A series of tests were ordered, including blood tests and a muscle biopsy.

Two weeks later, I received a call from the pediatrician's office, requesting my husband and I come to his office. It was one of the worst days of my life. We were told three things: our son had a rare, fatal disease called Pompe's Disease, there was no treatment available, not even an experimental drug, and the life expectancy for children with this disease was 6 months to one year. Ryan had just turned 6 ½ months old, and would probably not make it to Christmas four months away.

I was absolutely devastated. We were told to take him home and value the time we had left with him. Ryan did make it to Christmas, but he died one week before his first birthday. I cried every day for a long, long time.

Five years later, I was terrified to find myself expecting. I had a 25% chance of having another child with Pompe's Disease. But, I so desperately wanted another child. We decided to do prenatal testing to see if the baby would have the disease. When I was five months pregnant, we learned the test was positive. I was stunned; I was sure it couldn't happen again, even though I knew the odds. We had decided we would continue the pregnancy, regardless of the result, and love this child for however long we had him. But, the tears began again.

"Most parents find out they are carriers of Pompe's Disease, just like we did, when their babies are critically ill. By then, the disease was too far advanced, and their children were not healthy enough to be accepted into the trial."

We learned that Duke University was about to start a clinical trial with an experimental drug, but only had room for three babies. We watched as the spots filled. First one baby, and then the second, and I still had another month until delivery. It was heart-wrenching; I knew what would happen if we could not get into the trial.

In a horrible twist, we had an advantage. Most parents find out they are carriers of Pompe's Disease, just like we did, when their babies are critically ill. By then, the disease was too far advanced, and their children were not healthy enough to be accepted into the trial. Jason was born July 16th, and accepted into the trial as the third baby shortly after. He received his first infusion of the drug at 2 ½ months of age.

Jason is now a freshman at Iowa State University, majoring in Aerospace Engineering. He has mobility issues, but he lives a very independent life. He drives a car, has a part-time job, and is the President of his Residence Hall. He continues with weekly infusions, which are done in his dorm room.

We know that Jason is alive today because of Ryan. The harsh truth is that Ryan's death saved Jason's life. When Ryan was born, Pompe's Disease could not be detected through a blood test. It was only diagnosed months later, after the babies' muscles were damaged beyond repair. Caught early enough, the medications on the market today allow the children to live normal lives. I attend annual patient meetings at Duke University for Pompe's Disease infants. You can immediately tell which children received the medication early, and which did not. While I thoroughly enjoy seeing some kids running through the room, my heart breaks for those who cannot. I especially have compassion for their parents, as I know the guilt that a parent feels for not protecting their child.

Please, as a mother who has had two children affected by this terrible disease, I beg you to include Pompe's Disease on the newborn screening. Spare other families the hurt and loss that occurs without early notification.

Culture and Access to Services

Azeez Butali, D.D.S, Ph.D

Dentist, Associate Professor, Geneticist, Director of AfriCRAN and CEO of Healthcare Trends

"Access to care is as important as information about care. Some families may have limited access to care due to socio-economic situations or location (with little or no means of commuting).

"…the economic situation and cost of treatment will be deciding factors and in most cases jeopardize the chances of the child getting continuous care.

"[For my family] the cost of multiple admissions in the hospital, medications, time she missed school and other family lost hours was higher than the cost of a BMT [bone marrow transplant]."

You work with families who have difficulty understanding and accessing medical care for their children. What is your advice about how to improve communication and access to care throughout different cultures?

Communication and information are important for families to continue treatment or care. This is even more important for families who speak a minority language. For instance if a family feels they do not understand the need for continuous care due to a lack of communication, they may decide to ignore future appointments especially when the importance of such visit has not been understood.

Access to care is as important as information about care. Some families may have limited access to care due to socio- economic situations or location (with little or no means of commuting).

Again, regardless of whether the care or treatment is important, the economic situation and cost of treatment will be deciding factors and in most cases jeopardize the chances of the child getting continuous care.

Your family has experience with an inherited condition and Newborn Screening. What can you share about the treatment and your experience?

As a parent of a child who had sickle cell disease and received a bone marrow transplant (BMT) treatment 17 months ago, I can confidently state that it was a game changer. The treatment for sickle cell disease is the same as the treatment for X-ALD and MPS-I. It is a stem cell or bone marrow transplant.

My daughter is now looking healthier, her lungs and kidney that showed signs of damage are rapidly getting better, and her blood/oxygen levels are improving. She now breathes well and does physical activities that had been impossible for her before the transplant.

She has not been hospitalized since the transplant. This is a significant change for our family. She was diagnosed at 1 year old and was admitted to the hospital over 30 times from when she was 1 until she had the BMT. Hospital stays go from a few days to about 3 weeks each time. She was on multiple medications. Her quality of life was below average since she missed school and was limited as to the type of physical activities she could do. She was also prone to catching any infection around (survived swine flu in 2009).

Overall, the cost of multiple admissions in the hospital; medications, time she missed school and other family lost hours is higher than the cost of a BMT.

Where BMT is an option, I will opt for it. With growing scientific discoveries like CRISPR and others; the cost of treatment as well as BMT will be within reach.

Do you think it is important to add new conditions to the Newborn Screening Panel?

The decision to screen or not to screen depends on our need to learn and act towards preventing future children from being born with the diseases. When we screen, we are able to detect new cases and understand the pathogenesis of the diseases. The knowledge gained from the clinical presentations will drive research towards cheaper treatment and prevention, despite the fact that some of the conditions are late onset.

As a human geneticist, I view every opportunity for sample collection / screening as an opportunity to learn and drive science of discovery towards healthy population.

How do we educate parents if we do not know enough about it?

Will screening stop the child from living? The answer is “NO”

Will the parents be well informed of the likely course of child’s growth, development and quality of life? “Yes”

Will the information affect their lives? “Yes”

While the argument to only screen for conditions for which we have treatment for appears to make economic sense, it also limits our drive for scientific discoveries that will promote cheaper treatment and interventions.

Public Transportation in Iowa

Jeremy Johnson-Miller

Transit Programs Administrator, Statewide Mobility Coordinator

"This service operates within all of Iowa's 99 counties; however, we know not all medical services can be accessed within those counties or individual cities.

"[Medical Transportation] is a struggle when trying to make those connections for persons with extreme situations such as on-going treatments several hours away.

"Longer distances could mean higher cost and longer hours for traveling."

What is Public Transit in Iowa?

Public Transit has a service to provide transportation assistance for medical appointments. This service operates within all of Iowa's 99 counties; however, we know not all medical services can be accessed within those counties or individual cities.

We have a history of providing long distance medical shuttles to specialists in neighboring towns, crossing county lines to the University of Iowa Hospitals and Clinics, or even state lines to Mayo Clinic in Rochester, Minnesota.

What are some challenges for medical transportation?

While some transit systems are addressing healthcare access, many are not. This is a struggle when trying to make those connections for persons with extreme situations such as on-going treatments several hours away. In this case, we would need to work directly with that individual and the transit agency to determine cost and/or reliability of such a service.

How is this service paid?

Most of our services are out of pocket, which means the rider pays for the transportation. Many medical trips can also be covered by Medicaid, through Iowa's MCO providers. Our services are also reimbursed by federal and state funds.

Is this service available to families who travel to Iowa City for Newborn Screening appointments?

Public Transit in Iowa is always available to everyone and does not discriminate. Adding a new condition to Iowa's Newborn Screening Panel would not affect our daily work.

However, these appointments could complicate travel time and cost of services. Funding is limited, so we must charge the customer a fee. Longer distances could mean higher cost and longer hours for traveling. The average in-town trip costs $15, however the cost to the rider is minimal. The longer the distance, the costlier the service and more of a fee to be charged.

How can someone contact the Public Transit service?

We are constantly searching for new and innovative ways to deliver public transit services in Iowa, and welcome input from all citizens and various human service groups. Something unique to Iowa, is our network of Mobility Coordinators, who can be described as social workers for transit. These individuals work with community partners to locate gaps in service, working one-on-one with customers to determine new ways to get someone from point A to point B.

Public Transit in Iowa: www.iowadot.gov/transit
Mobility Management: www.iowadot.gov/iowamobilitymanagement

Newborn Screening Laboratory Services

Travis Henry, Ph.D

Laboratory Scientist,
State Hygienic Laboratory,
University of Iowa

"There is data from other states screening for these disorders where they have not identified any newborn cases. They have identified positive cases, but none of them have developed symptoms as a newborn.

"This means the effort spent screening for these disorders by the lab, clinical follow up, and administration has not provided the intended benefit

"No, I don't think it's a good idea to add these conditions to the panel at this time. As of now, it is not possible to tell the difference between newborn and late onset types"

What do you do for the Iowa Newborn Screening Program?

I review, assess, and implement the laboratory tests for newborn screening. I help the laboratory develop the screening for current and new disorders. I enjoy the opportunity to learn new information about the disorder and the underlying biology as well as the nuts and bolts of building a lab test that performs well and provides the information the program needs to help Iowans.

What do you think about these new disorders (Pompe Disease, MPS-I and X-ALD)?

The new disorders raise challenges for the Program that relate to how well the test works. There are labs already testing for these disorders, and the tests they have in place work very well. Our challenge is trying to figure out if these lab tests meet the goals of the Newborn Screening Program.

The new conditions are different from the other disorders for which the State screens because they all have a known late-onset presentation. This means these disorders may affect a baby in the newborn period, but these disorders may also not affect someone until they are children, teenagers, or adults.

There is data from States screening for these disorders where they have not identified any newborn cases. They have identified positive cases, but none of them have developed symptoms as a newborn. This means the effort spent screening for these disorders by the lab, clinical follow up, and administration has not provided the intended benefit.

It's difficult for the State to intervene in a resident's life and say we must test you for these disorders if they cannot demonstrate the benefit that was intended from the intervention.

How much would it cost to screen for the three New conditions?

The exact increase in cost has not been determined yet. The laboratory cost may seem relatively low, but the social/emotional cost to the family, and the future medical system would be high.

When Iowa starts screening for a new condition, they determine the cost of screening by doing pilot tests and experimenting with laboratory measurements. Then the program adds the cost of managing the screening and the follow up services to determine the final cost.

The cost of screening does not include the medical costs to Iowa residents and insurance companies for those cases identified as positive. With these new conditions, it may be difficult to predict when symptoms will occur, so there will be considerable costs for monitoring patients without symptoms who were identified by the screening.

How does this affect people who live in Iowa?

Since the testing typically cannot tell the difference between newborn and late-onset forms, there are Iowa residents who will be identified with a disease, but it may not be possible to tell them when symptoms will occur.

I think the Program has the responsibility to be able to tell residents with a high degree of certainty what their future will be like when it comes to the positive screen result. The State has intervened in their life without their choice and must assume responsibility for that action.

How does this affect the Newborn Screening Program?

The desire to help as many people as possible can overwhelm the people receiving the service if the service doesn't truly meet their needs. Screening for the new disorders may not be meeting the needs of the community if it cannot provide relevant information, and help improve the lives of newborns.

Newborn screening improves the lives of babies by identifying those at risk and providing an opportunity for early intervention. Newborn screening provides an important service. The screening doesn't meet the goals of the program if it doesn't improve the child's life or offer opportunities for early intervention.

Do you think it's a good idea to add these conditions to the panel? Why or why not?

No, I don't think it's a good idea to add these conditions to the panel at this time. As of now, it is not possible to tell the difference between newborn and late onset types. Perhaps with additional information, research, and use of new technology, it may be possible to tell newborn from late onset.

Because screening for Pompe disease, MPS-I, and X-ALD differ for newborn or late onset, adding the conditions to Newborn Screening seems to be population research. Collecting information about a disorder and using the information to make improvements to laboratory test should only happen with voluntary participation. It should not happen through a mandatory State Newborn Screening. Iowa residents should decide if they want to participate in research, not have the State decide for you.

Newborn Screening Follow-up Program
University of Iowa Stead Family Children's Hospital

Carol Johnson

Supervisor,
Newborn Screening Follow-up,
University of Iowa

"Adding these new conditions to the Iowa Newborn Screening panel would mean that the program would need to hire at least one more genetic counselor and potentially another follow up nurse…we would need to establish some sort of long term follow up system, which we do not currently have… I'm not sure there would be the funding necessary to add this component to the NBS.

"I would be in favor of adding these new conditions if the screening only detected the infantile forms of these conditions and if the false positive rate was lower."

Who are you and what do you do for the Iowa Newborn Screening Program?

I am the supervisor for the follow up services of the Newborn Screening program at the University of Iowa. The laboratory notifies us when they find a positive screen for any of the conditions on the Newborn Screening panel. Our staff calls the baby's pediatrician or family doctor, and makes sure the baby sees the doctor right away. The baby works with the specialists to get a diagnosis and medical services.

What does your program need to start screening for these three conditions (Pompe disease, MPS-I, and X-ALD)?

Adding these new conditions to the Iowa Newborn Screening panel would mean that the program would need to hire at least one more genetic counselor and potentially another follow up nurse as well.

Since these conditions have the potential to be late onset (ie not present at birth, but could be diagnosed in people up to age 50 or later), we would need to establish some sort of long term follow up system, which we do not currently have. It would be a significant investment of time and money to get this type of follow up established.

I'm not sure there would be the funding necessary to add this component to the NBS. Adding more staff and a long term follow up program would ultimately increase the cost of screening for all babies born in Iowa.

How would screening for these new conditions affect people who live in Iowa?

Whenever you can screen for disorders it benefits those babies and families who have the disorders. Often, getting a baby to treatment sooner can mean improved health, less disability, and even can save a baby's life. You can't put a price on a person's life or on their quality of life.

However, with any disorder that is screened for, there will always be false positives. A false positive means that the screen detected an increased risk of a particular disorder for a baby that needs to be investigated further. This investigation could include getting another screen, having blood drawn for further testing, a visit to a specialist, or other interventions. False positives can also cause increased anxiety in parents of a baby with an abnormal screen.

Newborn screening programs need to weigh the benefit of identifying babies with disorders against the number of false positives that there will be and the impact to those families whose babies ultimately do not have the disorder. There will always be more false positives than true cases no matter how well the screening process is performed.

How does this decision to add new conditions impact the Newborn Screening program?

The newborn screening community cares deeply about every baby born in our state and feels a strong sense of responsibility to the baby and their family. While adding these new conditions is a big win if you can identify a newborn with a condition, does that win have the same positive impact when it's the late onset form of the disorder? These are some of the moral and ethical issues newborn screening programs struggle with.

In addition, if these disorders were added to the panel without proper funding, staffing, programming, specialists, and infrastructure it could prove disastrous to the program and to the families we serve.

Do you think it's a good idea to add these conditions to the panel? Why or why not?

I would be in favor of adding these new conditions if the screening only detected the infantile forms of these conditions and if the false positive rate was lower.

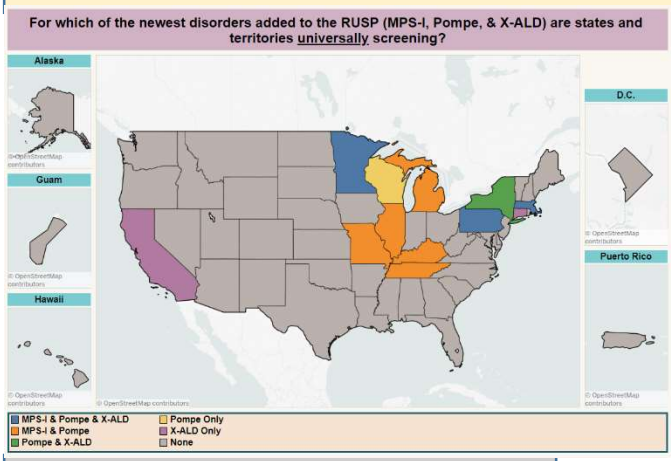
Screening for disorders that have a late onset is beyond the scope of newborn screening – remember – the name of the program is <u>newborn</u> screening.

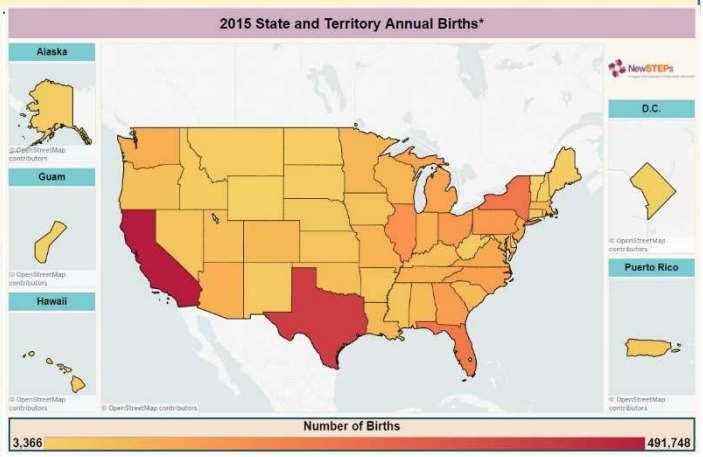
If families are told that their baby has the late onset form of these conditions, will they truly understand what that means? Will they tell their child that they have this condition? Will that information be transferred to a new health care provider, particularly when a child transitions from pediatric to adult care? What kind of worry are we setting up for parents and patients to go through? Will they always wonder if today be the day that they will show symptoms?

One of the benefits listed for screening for these disorders is to reduce/eliminate the long diagnostic journey that many patients with these disorders experience. I'm not sure it will, as my long experience in health care tells me that health information is often lost or transmitted incorrectly – like the game of telephone.

Will people truly experience benefit from screening for these disorders if it is the late onset form? Yes – if we find the infantile form. I would say no if we find the late onset disorders or if we can't reduce the false positive rates for some of these disorders.

Other States' Experiences: Missouri and New York





Patrick Hopkins
State Public Health Laboratory
Jefferson City, Missouri

Number of years screening:
Pompe disease: 5 years
MPS-I: 5 years

Beth Vogel
Wadsworth Center,
NYS Department of Health
Albany, New York

Number of years screening:
X-ALD: 4 years
Pompe disease: 3.5 years

-Missouri-

How long have you been screening for these New Conditions?

We've been screening for Pompe disease and MPS-1 since January 11, 2013 (5 years). We are waiting on a new laboratory kit to be developed before starting screening for X-ALD.

How many true positives (diagnosis) have you found?

We were one of the first states to begin screening so our numbers are higher. We have found 36 true positive Pompe disease screenings out of 224 screen positive results. We have found 2 MPS-I out of 190 screen positive results.

How many unclear results (false positives or variants of unknown significance) have been found?

With Pompe disease we have found 7 Conditions of Unknown Significance or Unknown Onset, 44 Pseudo-deficiencies, 53 Carriers, 4 lost to follow up, and 10 that are currently pending.

With MPS-1, we have found 4 Conditions of Unknown Significance or Unknown Onset, 87 Pseudo-deficiencies, 12 Carriers, 6 lost to follow up, and 17 that are currently pending.

How long does it take to determine a diagnosis after the initial results come back?

The additional tests can take up to 2 weeks. If infantile Pompe disease is suspected, a heart abnormality can be detected with initial clinical evaluation of the baby. Treatment with Enzyme Replacement Therapy would not begin until the DNA sequencing results have complete diagnosis.

Have you been able to determine early onset (before 1 year) vs. childhood onset vs adult onset for these New Conditions?

So far yes, for early onset, but we cannot determine childhood onset vs adult onset. The childhood and adult onset are a spectrum and depend on many other factors, some we are not yet aware of.

-New York-

How long have you been screening for these New Conditions?

We have been screening for X-ALD since December 30, 2013 and Pompe disease since October 30, 2014. We have screened 1,101,027 babies for X-ALD and 901,435 babies for Pompe disease.

How many true positives (diagnosis) have you found? How many unclear results (false positives or variants of unknown significance) have you found?

For X-ALD we have reported 73 patients:

- 26 of these patients were confirmed with diagnosis,
- 23 were carriers,
- 8 were Zellweger diagnosis (a similar, more severe, and untreatable condition that results in death before 1 year of age), and
- 16 who were non X-ALD cases: Aicardi-Goutieres Syndrome (https://bit.ly/2FUnxjB), Variant of unknown significance, D-Bifunctional Protein Deficiency

For Pompe disease we have reported 118 patients:

- 5 confirmed cases of infantile Pompe disease,
- 56 carriers (35 with pseudodeficiency),
- 45 possible late-onset (22 of them considered probable) and
- 12 with unknown significance.

How long does it take to determine a final diagnosis after receiving the initial results?

For X-ALD we do additional tests within the laboratory and get the results back within about a week. For Pompe disease, the additional tests also take about a week, but there are cases that aren't clear and may require monitoring over time. These could take much longer to get a final diagnosis

Have you been able to determine early onset (before 1 year) vs. childhood onset vs adult onset for these New Conditions?

Differentiating the age when symptoms appear isn't possible for the X-ALD condition. Pompe disease is easy to determine infantile forms of the condition.

Adding Conditions to State Newborn Screening Panels: Recommendations for Decision Makers

Newborn screening is the practice of screening every baby prior to hospital discharge for certain harmful or potentially fatal metabolic and genetic conditions that are not otherwise apparent at birth. Newborn screening allows babies to be identified and treated before they get sick, preventing serious health problems or even death. It is the largest and most successful health promotion and disease prevention system in the country, and the fastest, safest way to help protect against certain diseases and medical conditions.

KEY CONSIDERATIONS

Though technology has made it possible to screen for an increasing number of conditions, evidence does not support screening for all detectable disorders. Decision makers should consider multiple factors to assess the value and feasibility of adding a condition/s to the state's newborn screening panel. These include:

- Is there sufficient evidence of the effectiveness of screening for the condition?
- Will the child and family benefit from early detection?
- How would addition of this condition align with other state policies and values?
- Is funding available to support all costs of implementation including parental education, follow-up, diagnosis, treatment and management, and program evaluation?
- Do the responsible state agencies have the staffing and administrative capacity to support implementation of a new condition?

States should evaluate addition of a condition/s to the state's screening panel using established state or federal deliberative processes that apply recommended criteria and obtain input from relevant stakeholders. Where feasible, states should conduct pilot studies on potential conditions and provide a process for parental consent.

SPEAKING WITH STATE DECISION MAKERS

Newborn screening specialists and their supporters can frame discussions with state decision makers using the following recommended criteria for adding conditions:

- Universal screening is critical to identify all babies who may need treatment
- Affected babies will have a significant, life-challenging risk of illness, disability or death if not treated in the newborn period

- Effective treatment is available for the condition/s
- Treatment during the newborn period is more beneficial than later treatment
- Resources for and access to treatment and counseling are universally available
- The health and societal benefits outweigh the risks and burdens of screening and treatment on newborns and relatives.

LIMITING THE UNINTENDED CONSEQUENCES OF LEGISLATION

Sometimes, despite the best efforts of newborn screening specialists and supporters, legislation will advance that is counter to their recommendations. In these instances, newborn screening allies should advocate that the bill include:

- Sufficient funding for time and costs of implementation as well as administration
- Parental consent required for conditions added in the absence of screening recommendations through an established national or state deliberative process
- Rigorous evaluation required at set intervals to measure the effectiveness of population-wide screening for the condition/s
- Strong protection from liability for screening of the condition/s.

ACKNOWLEDGMENTS / SOURCES

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fact sheet

march of dimes
A FIGHTING CHANCE FOR EVERY BABY
Iowa

Support Adding Pompe, X-ALD, & MPS I to the IA Newborn Screening Panel

The March of Dimes supports conditions added to the state newborn screening panel only when they are included on the Recommended Uniform Screening Panel (RUSP).

What are Pompe Disease, X-ALD, and MPS I?

Pompe disease is a rare, inherited disorder that causes progressive muscle weakness. It is a part of a group of disorders referred to as Lysosomal Storage Disorders (LSDs) and is classified as either infantile- or late-onset disease. X-linked Adrenoleukodystrophy (X-ALD) is a rare, inherited disorder that causes damage to the nervous system and kidneys. It is one of a group of genetic disorders called leukodystrophies. Mucopolysaccharidosis Type 1 (MPS I) is an inherited disorder that can affect many parts of the body. It is also classified as a LSD like Pompe disease.

Why Screen for Pompe, X-ALD, and MPS I?

Pompe disease, X-ALD, and MPS I can all be detected using the traditional newborn dried bloodspot. Research has shown that early treatment and management can lead to better outcomes for those diagnosed with infantile-onset Pompe disease. Infants with X-ALD or MPS I often appear healthy at birth and the condition may go unnoticed during a critical period without newborn screening. Research has shown that treatment prior to the onset of symptoms can be lifesaving or lead to better quality of life for those with X-ALD. There is no cure for MPS I, but treatment can delay the progression, prevent or reduce permanent tissue and organ damage, and improve the quality of life

All Newborns Should Be Screened for Pompe Disease. X-ALD, and MPS I

The March of Dimes supports screening all newborns for conditions placed on the RUSP by the U.S. Secretary of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC). The RUSP includes conditions for which there is a documented medical benefit to the affected infant from early detection and treatment; there is a reliable screening test for the disorder; and early detection can be made from newborn bloodspots or other means. Pompe disease, X-ALD, and MPS I are all included on the RUSP.

Key Points

- Newborn Screening (NBS) is a public health program the provides early identification and follow-up for treatment of infants affected by certain genetic, metabolic, hormonal, and/or functional conditions.
- Currently, each state or territory operates, by law, its own NBS program.
- 34 core disorders are listed on the Recommended Uniform Screening Panel (RUSP) by the U.S. Secretary of Health and Human Services.
- IA has added 31 of the 34 core disorders on the RUSP to the state NBS panel.
- The March of Dimes supports the addition of conditions to state newborn screening panels **ONLY** when they meet the three criteria **AND** have been added to the RUSP.
- March of Dimes is also supportive of state policies that include a provision that would automatically add conditions to the state newborn panel once it is approved for the RUSP.

Contact information:
Matt Keppler, Director of Advocacy and Government Affairs, at mkeppler@marchofdimes.org or (314) 306-5310

The March of Dimes is a national voluntary health agency whose volunteers and staff work to improve the health of infants and children by preventing birth defects, premature birth and infant mortality. Founded in 1938, the March of Dimes funds programs of research, community services, education and advocacy. For the latest resources and information, visit ***marchofdimes.org*** or ***nacersano.org***.

fact sheet

march of dimes
A FIGHTING CHANCE FOR EVERY BABY
Iowa

The Process of Newborn Screening

Newborn Screening

Newborn Screening (NBS) is a public health program that provides early identification and follow-up for treatment of infants affected by certain genetic, metabolic, hormonal, and/or functional conditions. The success of NBS programs has made screening routine for the over four million infants born in the United States each year. Screening tests are done using a few drops of blood from a newborn's heel, usually taken in the hospital 24- 48 hours after birth, or by functional testing such as a hearing test, and pulse oximetry test.

Currently, each state or territory operates by law, it's own NBS program. Although all states have laws that require screening, individual programs vary in the number and types of condition for which newborns are screened. In 2000, the March of Dimes led the way in proposing a national standard for newborn screening which included a core list of 9 disorders, with provision for expanding the list as science and technology evolved. In August 2004, the American College of Medical Genetics (ACMG) submitted a report to the federal Health Resources and Services Administration (HRSA) which included proposed nationwide standards for state newborn screening programs. The report listed 29 core treatable disorders that should be targeted by newborn screening programs and an additional 25 secondary target conditions for which test results should be reported. As of January 2017, there are now 34 core treatable disorders and 27 secondary target conditions. These secondary target disorders are not actively sought by newborn screening, because they do not yet have documented treatments or there is limited knowledge of their natural history.

Adding Newborn Screenings to the State Panel

The March of Dimes played a vital role in the passage of federal legislation which supported the creation of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) and established the Recommended Uniform Screening Panel (RUSP). Currently, conditions are added to the RUSP upon approval by the U.S. Secretary of Health and Human Services, based on evidence review and a recommendation from the SACHDNC.

The March of Dimes supports the addition of conditions to state newborn screening panels only when they meet the following three criteria: 1) there is documented medical benefit to the affected infant from early detection, and treatment; 2) there is a reliable screening test for the disorder; and 3) early detection can be made from newborn blood spots, or other specific means. Conditions added to the RUSP must meet the three March of Dimes criteria for adding new newborn screening conditions. March of Dimes encourages states to provide education to parents about conditions that are not yet on the RUSP.

The March of Dimes is supportive of state policies that include a provision that would automatically add conditions to the state newborn panel once it is approved for the RUSP by the U.S. Secretary of Health and Human Services.

Key Points

- Newborn Screening (NBS) is a public health program the provides early identification and follow-up for treatment of infants affected by certain genetic, metabolic, hormonal, and/or functional conditions.
- Currently, each state or territory operates by law, is own NBS program.
- 34 core disorders are listed on the Recommended Uniform Screening Panel (RUSP) by the U.S. Secretary of Health and Human Services.
- Approximately 40,000 babies are screened for treatable conditions in Iowa every year.
- The March of Dimes supports the addition of conditions to state newborn screening panels **ONLY** when they meet three criteria **AND** have been added to the RUSP.
- The March of Dimes is also supportive of state policies that include a provision that would automatically add conditions to the state newborn panel once it is approved for RUSP.

Contact information:
Matt Keppler, Director of Advocacy & Government Affairs, at mkeppler@marchofdimes.org or (314) 306-5310

Local Pediatricians

Gretchen Vigil, MD
Pediatrician,
University of Iowa Hospitals and Clinics,
Iowa City

Sarah Wickenkamp, MD
Pediatrician,
Stead Family Children's Hospital, Cedar Rapids

Daun Pringle,
Laboratorian,
Mary Greeley Medical Center
Ames

Marianka Pille, MD
Pediatrician
Des Moines University Clinic, Unity Point Health, Blank Children's Hospital
Des Moines

What do you think about Iowa adding new conditions to the Iowa Newborn Screening Panel (Pompe disease, MPS-I, and X-ALD)?
I do not have any patients with these conditions. (I do have a scattered handful of those affected by current screening program.)

I think the most helpful information would be the reliability of the test and how great the chance of false positives. I think some of the issues raised won't be as important after it gets rolled out.

Maybe there is no good current treatments, but there can be increased monitoring, increased awareness, and decreased delay of diagnosis.

If the false positive rate is higher than desired, I wouldn't do the screening due to cost in dollars and emotional stress.

-Gretchen Vigil, Pediatrician, University of Iowa Hospitals and Clinics, Iowa City

How would adding these new conditions affect your work as a pediatrician?
As with everything else in medicine, it would likely increase my workload since I would have to be able to explain these conditions to parents, especially false positives.

Do you think we should add new conditions to the Newborn Screening Panel?
No. Just because we CAN test for something doesn't mean we should. I think we are already having issues w/ fair division of the health care dollars available.

-Sarah Wickenkamp, Pediatrician, Stead Family Children's Hospital, Cedar Rapids

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Do you think we should add new conditions to the Newborn Screening Panel?
I do not see the point in adding a very expensive test to the screening battery already in place, especially if it has a high rate of false positives. This means calling patients back, and that always creates a lot of stress for the parents. Many of our parents travel quite some distance to see a pediatrician, and to have that NBS repeated would be quite the ordeal for them.

What are some reasons why families follow-up with treatment?
The lack of care availability and cost is definitely an issue, for a multitude of different diseases and patient groups. Through my interactions with other tests, and hearing their trials and tribulations (along with my own personal medical issues): cost and how far I have to drive is definitely considered into my compliance. A far distance means missing work, which for many this means possibly losing their jobs - which could be the source of their insurance.

Do the families you work with understand the Newborn Screening program?
Families don't understand the newborn screening, other than it's state mandated test. They know it screens for a bunch of things, but they don't understand the urgency or how severe those diseases could be. When we notify the physician that a recollection is necessary, often times the nursing staff doesn't explain why it has to be done. That gets left to us in the laboratory when they come from the recollection. If it's a poor quality specimen, they want to know why the 5 big circles weren't enough. They don't understand the timing of having to be at 24 hours, etc. For being a really simple collection. Our parents who understand it the best are the ones with other children who have been saved by these! Genetics does such an amazing job of preparing families for their second babies!

-Daun Pringle, Laboratorian, Mary Greeley Medical Center, Ames

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How would adding these conditions to the panel affect your work?
Overall I would not anticipate dramatic shift in my work, as these are rare conditions and presumably abnormal results of screening would be infrequent. I would anticipate occasional calls/faxes from the newborn screening program, which in turn add time to office visits to discuss results, and potentially require phone calls etc to coordinate care. I would expect this to be a small overall time commitment.

How would it affect the people you serve?
My population is ethnically diverse therefore lots of opportunity for rare inherited disorders, but even so I do not have many patients with conditions that are screened for (abnormal results are rare). For those with true positive abnormals, early diagnosis results in early intervention and better health and long term outcomes. It can prevent devastating health crises. For those with false positive abnormals, the screen would add stress for parents as they wait for confirmatory testing, and would add procedures (blood draw usually) for the patient.

How would it affect your community?
I would expect a very small number to be affected. Since everyone is already screened, there are no new processes to incorporate. For those who have a diagnosis made early due to screening (a small number) there would be significant and lasting impact. For those who have an abnormal result, but ultimately no diagnosis (also hopefully a small number) there would be a short interval of anxiety/worry hopefully without lasting impact.

Does the cost of treatment and access to treatment affect how many of your patients follow through with recommended medical treatment?
This is an undeniable issue. Transportation in particular is difficult since many of the metabolic specialists are in Iowa City or Rochester and I am in Des Moines. That said, there are ways to access resources and facilitate complex care, and to the extent that we can change outcomes by screening and starting treatment early for rare congenital diseases we may be able to decrease the need for these resources

Do families understand the screening and the results?
For the most part, yes. Sometimes it takes longer to explain but ultimately, I think I and other pediatricians are able to clarify the reason for the test, the need for any retesting, and the management of any issues identified

Do you think it's a good idea to add these diseases to the panel? Why or why not?
Yes I do think it is a good idea. I am not an expert on these diseases, but my understanding of newborn screening is that it allows interventions and treatments to be provided early, before symptoms begin or damage becomes permanent. Trusting that the diseases under consideration have such interventions available, it makes sense to add the testing since for the newborn it would not change the fact that they are having screening done. All babies already have blood collected and this would not change. Things would only change for the few true positives (presumably a helpful intervention and improved health) and the false positives (negative impact of stress and extra lab tests). On balance this favors the testing in my opinion.

-Marianka Pille, MD
Des Moines University Clinic, Unity Point Health, Blank Children's Hospital

Public Health Department

Marcus Johnson-Miller

Bureau of Family Health
Iowa Department of Public Health

What does the Iowa Newborn Screening Program do and how is it related to Public Health?

In May 2011, the Centers for Disease Control and Prevention (CDC) named newborn screening one of the greatest public health achievements of the 20th Century. For over 50 years, newborn screening programs nationwide have screened over 4 million babies each year for inherited diseases. In Iowa, about 39,000 babies are screened each year for hearing loss, congenital heart disease, and certain genetic diseases.

The US Secretary of Health and Human Services Advisory Committee for Heritable Disorders in Newborns and Children (ACHDNC) developed a Recommended Uniform Screening Panel (RUSP), which list all of the conditions recommended for state newborn screening panels (meaning every state is recommended to screen for the conditions on the RUSP). The Iowa Newborn Screening Program (INSP) currently screens for 31 of the 34 core conditions on the RUSP.

What do you think about Iowa adding new conditions to the Iowa Newborn Screening Panel (Pompe disease, MPS-I, and X-ALD)?

As we examine new conditions for addition to Iowa's newborn screening panel, we must consider several things in order to continue our successful program.

The Iowa Newborn Screening Program at the Iowa Department of Public Health considers the RUSP to be a recommendation, just as the name implies.

"It is the responsibility of the IDPH to assure that its mandated newborn screening program

1. **meets the needs of the Iowa population,**
2. **has the equipment and staff to do the testing,**
3. **has in place trained, specialized medical staff with expertise in treating newborns with the conditions,**
4. **has funding available to conduct pilot screening for each new condition before it's added to the Iowa screening panel,**
5. **each condition has passed a rigorous, evidence-based review and has been approved to the panel by the Advisory Committee and the Iowa State Board of Health."**

It is the responsibility of the IDPH to assure that its mandated newborn screening program

1. meets the needs of the Iowa population,
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3. has in place trained, specialized medical staff with expertise in treating newborns with the conditions,
4. has funding available to conduct pilot screening for each new condition before it's added to the Iowa screening panel,
5. each condition has passed a rigorous, evidence-based review and has been approved to the panel by the Congenital and Inherited Disorders Advisory Committee and the Iowa State Board of Health.

In some states, the legislature has passed a law requiring the state to screen for certain conditions. Having the newborn screening panel determined by state law poses problems: the capacity of the newborn screening program to screen for the conditions may be lacking, and the newborn screening program may not be able to meet the timelines set forth by the law, and there might not be treatments readily available in the state, amongst other issues.

The Iowa Congenital and Inherited Disorders Advisory Committee has a well-established procedure in place for the evidence-based review process for new conditions. This process allows the INSP to be in the best position to continue its life-saving work.

Legal Considerations

What do you think about Iowa adding new conditions to the Iowa Newborn Screening Panel (Pompe disease, MPS-I, and X-ALD)?

Iowa Law: General Purpose

For over forty years, Iowa's Legislature has directed the Iowa Department of Public Health to conduct a newborn screening program to detect congenital and inherited disorders (formerly called birth defects). The legal purpose of the program is "to reduce and avoid adverse health conditions" of Iowans.[1]

To achieve this purpose, the Legislature has established within the Department a Center for Congenital and Inherited Disorders, which performs the following duties: (1) conducts and supervises the screening program for disorders amenable to population screening, (2) conducts and supervises other health programs to aid in early detection, treatment, prevention, education, and provision of supportive care related to these disorders; (3) gathers and maintains information regarding disorders; (4) monitors these disorders to determine occurrence and trends and to assist in planning services to children and their families; (5) implements public and health education programs; and (6) participates in policy development.[2]

Screening Requirement

Iowa law provides that "all newborns in this state shall be screened for congenital and inherited disorders in accordance with rules adopted by the department." Attending health care providers have a duty to "ensure that every newborn under the provider's care is screened for congenital and inherited disorders in accordance with the rules adopted by the department."[3]

Heather Adams

State Assistant Attorney General
Office of the Attorney General
of Iowa

"In sum, newborn screening raises legal issues which are present in many discussions about public health issues: namely, how do we protect the public's health while respecting individual and family rights and choices?

"The balancing of these interests requires weighing the medical and scientific facts available, considering risks and benefits both to society and to individuals, and informing and educating decision-makers and parents to the greatest degree possible."

[1] Iowa Code § 136A.1
[2] Iowa Code § 136A.3
[3] Iowa Code § 136A.5

Confidentiality

The Center and the Department are required to maintain the confidentiality of all information which identifies a child or parent involved in newborn screening.[1] This information is carefully protected from disclosure, as are the newborn screening specimens.[2]

Parental Refusal

Iowa law does allow a parent to refuse the screening. If a parent objects to the screening the health care provider must document the refusal in the baby's medical record, obtain a written refusal from the parent, and report the refusal to the Department.[3] The written refusal form informs the parent that if certain conditions are undetected and untreated they may cause permanent damage, including intellectual disabilities, growth failure, and death. If the parent refuses the screening, they accept the legal responsibility for the consequences of that decision.

Screening Panel

The 49 specific conditions included in the newborn screen are determined by the Department and approved by the State Board of Health and can be found here: http://idph.iowa.gov/Iowa-Newborn-Screening-Program/For-Providers-and-Professionals/Conditions-Screened.[4]

To decide whether to add a new disorder to the panel, the Department is required to follow protocols which involve nomination of the condition and a review of the condition by the Center's Newborn Screening Panel Management Subcommittee. The Subcommittee is required to consider certain criteria to determine whether to recommend adding a condition, including:

1. The condition under review should be an important health problem.
2. There should be an accepted treatment for patients with recognized disease.
3. Facilities for diagnosis and treatment should be available in Iowa or the region.
4. There should be a suitable test or examination and Iowa should have the capacity to develop and provide the test.
5. There should be an agreed policy on whom to treat as patients.
6. Financial feasibility. The cost of case finding should be economically balanced in relation to possible expenditure on medical care as a whole.
7. The potential impact of integrating the test on existing NBS protocols, programs, systems, and the Department.
8. Position statements from appropriate local and national organizations.

The results of this community engagement project will be presented to the Subcommittee to assist it in deciding whether to recommend adding these conditions to the screening panel.

Additional Legal Considerations

Prior to adding a new condition to the required screening panel, consideration should be given to the fact that the screening panel is a state-mandated requirement, and "the greater the problems with reliability, predictive value, or known treatments, the less basis there is for a state testing mandate."[5]

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[1] Iowa Code § 136A.7
[2] 641 IAC 4.3(7), 4.3(8)
[3] Iowa Code § 136A.5(3)
[4] Iowa Code § 136A.5(1); 641 IAC 4.3(1)
[5] Jennifer Kraszewski, *Legal Issues in Newborn Screening: Implications for Public Health Practice and Policy*, Public Health Reports, 2006 Jan-Feb, 121(1)

Additionally, when considering adding a condition for which the newborn screen may not yield results which are clearly predictive or result in defined immediate treatments, recommendations for educating and counseling parents about screening for these conditions should be discussed as well.

In sum, newborn screening raises legal issues which are present in many discussions about public health issues: namely, how do we protect the public's health while respecting individual and family rights and choices? The balancing of these interests requires weighing the medical and scientific facts available, considering risks and benefits both to society and to individuals, and informing and educating decision-makers and parents to the greatest degree possible.

Economic considerations

David Swenson

Associate Scientist
Department of Economics
Iowa State University

"If the benefits of an initiative are equal to or greater than the costs of providing those benefits over some reasonable period of time, then society is better off. If society is better off, then government, if it has the resources, has a justification for funding a course of action.

"The research to date does not provide findings that suggest that there are significant costs avoided, substantial total beneficial health outcomes, or reliable estimates of the costs of the recommendation to the public as well as to households in forms that would allow for a conventional benefit costs analysis"

Help me understand economics. How does the government decide what is in the public's best interest?
We try to be objective when we make important decisions. We weigh the pros and cons. We rank and prioritize. We compare our wants and needs to our abilities to pay. We consider how our decisions affect others. We also try to live within our means.

Governments, because they are acting in the public's interest, are charged with being both *efficient* and *effective* with regard to the choices they make and the policies they implement. Programs should attain agreed-upon objectives and do so at a reasonable costs.

When implementing new public programs, society often demands a higher standard for decision making. This higher standard is called *benefit-cost analysis* (BCA). BCA is a formalized approach to deciding the worth and utility of a project or proposal. In the simplest terms, if the benefits of an initiative are equal to or greater than the costs of providing those benefits over some reasonable period of time, then society is better off. If society is better off, then government, if it has the resources, has a justification for funding a course of action. Projects that produce comparatively more benefits over costs will be prioritized over others.

Is it different when programs are related to health?

When a regulation involves a public health requirement, BCA can become more complicated. In the face of epidemics or other public health emergencies, government often act without knowing either the full benefits or the full costs of action. Formal BCA rules are relaxed during emergency situations where significant individual and societal costs are implied by not acting and broad benefits are presumed.

One can see this by rapidly rising opioid addiction and death rates in the U.S. and the state-by-state and, of late, national clamor attempting to deal with this public health concern.

With incremental changes to public health requirements, however, such as mandatory vaccinations, recommendations for periodic examinations, or, in this case, neonatal screening for disease or disorder, promoters of those increments are obliged to use agreed-upon criteria to determine the benefits to the individuals, their families, and to society in light of all costs as well as harms that the changes might generate. Harms can take the forms of pain, suffering, discomfort, stigmatization, or risk of death associated with both diagnosis and treatment; cost burdens to families or governments; and the anxieties associated with incorrect diagnoses or false-positive test results, as examples.

What are some questions to ask when deciding on the benefits and costs of this decision?

Among the topics at hand, there are recommendations to include screening for Mucopolysaccharidosis Type I (MPS-I) for newborns. The presumption is that this addition to the screening panel for newborns would provide better health outcomes for those with this condition, or allow for earliest possible treatment. On the way to comparing the benefits and costs of this adding MPS-I, there are several questions that benefit-cost analysts would initially ask:

1. *Is the newborn screening an efficient mechanism for detecting MPS-I?* Answer: based on published and unpublished data, the experts expect nearly 31 false positive screens for every true positive result.

2. *Will the newborn screening produce a meaningful improvement in MPS-I identification?* Answer: When comparing expected number of cases identified at some later date based on symptoms with cases identified through newborn screening, the experts say that there would be the same number of severe cases detected in each, but that the newborn screening would result in a substantial number of unknown identifications. In all, a nationwide implementation of screening would yield 44 cases of confirmed or possible MPS-I, compared to an expected 40 cases using later symptom-based detection – four more cases nationwide out of 4 million births.

3. *Will the newborn screening produce significantly improved health outcomes?* Answer: The data are inconclusive, though improvements in having severe complications were indicated. The health experts projected that early detection *could* result in from 0 to 2 deaths avoided by 5 years of age, but the evidence of increased survivability is inconclusive.

4. *Are the costs of MPS-I screening known?* Answer: the initial cost increment to the existing protocols is considered very small. However, costs of program implementation, additional screening and follow-up tests for false positives or unknown types, along with staff education and training, would be high but specifically how much is not known.

5. *Are there measurable costs to households associated with the newborn screening?* Answer: owing to the very high rate of false positives and the apparent inability to diagnose the less severe types of this condition through the initial screening, there would be both anxiety and stress imposed upon families as well as substantial material costs associated with follow-up testing.

6. *Finally, is there sufficient and reliable research-based evidence that demonstrates clearly that the benefits to individuals and to society by including MPS-I as part of the newborn screening panel equal to or greater than the costs associated with implementing and maintaining this program?* Answer: The research to date does not provide findings that suggest that there are significant costs avoided, substantial total beneficial health outcomes, or reliable estimates of the costs of the recommendation to the public as well as to households in forms that would allow for a conventional benefit costs analysis.

Insurance Coverage

C. David Smith, MD, MA, FACS

Medical Director
Iowa Medicaid Enterprise,
hawk-i and
Clinical Advisory Committee

"With Iowa Medicaid Enterprise (IME) financing approximately 15,000 to 16,000 births per year, this could potentially expand Iowa Medicaid's financial commitment by more than $480,000 per year.

The IME clinical advisory committee did not support coverage for expanding the newborn screening panel to include these three diseases both because of the rarity of these conditions and the added cost of discovery."

What do you think about Iowa adding new conditions to the Iowa Newborn Screening Panel (Pompe disease, MPS-I, and X-ALD)?

The Secretary of the US Department of Health and Human Services has recommended the expansion of state newborn screening panels to include three rare genetic diseases, Pompe disease, Mucopolysaccharidosis I (MPSI) and X-linked Adrenoleukodystrophy (X-ALD). The cost of screening for these conditions is still under review. This cost would be expected to be covered by the insurer. With Iowa Medicaid Enterprise (IME) financing approximately 15,000 to 16,000 births per year, this could potentially expand Iowa Medicaid's financial commitment by more than $480,000 per year.

Discussions were held with both the ***hawk-I*** (Children's Health Insurance Program) clinical advisory committee and the Iowa Medicaid clinical advisory committee. The rationale for newborn screening of diseases focuses on early treatment to prevent irreversible complications which can be avoided or lessened with identifying those are risk.

At the current level of births financed by IME, which is close to 16,000, the newborn screening for these diseases could identify 1-2 infants with one of the three diseases each year. The IME clinical advisory committee did not support coverage for expanding the newborn screening panel to include these three diseases both because of the rarity of these conditions and the added cost of discovery.

In the State of Iowa where there are approximately 40,000 births per year, these genetic conditions would be anticipated to be found in 3-4 infants per year. These children frequently turn to IME for coverage of their expensive therapy which may be compromised by the added costs of screening. For these reasons, the added coverage was not recommended by either the ***hawk-i*** clinical advisory committee or IME clinical advisory committee.

**Side note: The Iowa Newborn Screening Program consulted Inherited Disease Specialists at the University of Iowa Hospital about current reimbursement with Iowa Medicaid for Pompe disease, X-ALD and MPS-1. They stated any treatment that is considered the standard of care and medically necessary is typically covered by an insurance provider. Some providers require pre-authorization, however, lately Iowa Medicaid has not required pre-authorization for treatments for Biotinidase Deficiency.*

Currently there are no patients with MPS-I at their clinic. There is one patient with Pompe disease and he/she does not have Medicaid insurance. This patient's insurance (BCBS) is covering the cost of Enzyme Replacement Therapy.

There are other patients with Gaucher, Fabry, and Hunter disease (similar treatment as MPS-Iand Pompe disease) and some have Iowa Medicaid insurance. Enzyme Replacement Therapy iscovered for all the patients. Enzyme Replacement Therapy is put out by 3 different pharmaceutical companies and only 1 is on the formulary and approved for reimbursement, which is similar to other brand-name drugs.

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